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(54) Title: HUMAN *FMO3* GENE MUTATIONS AND POLYMORPHISMS, AND USES THEREOF

(57) Abstract: The present invention relates to human *FMO3* gene polymorphisms and more particularly to uses thereof in the diagnosis of trimethylaminuria. There is provided a method for detecting an altered metabolism of a substrate of a flavin-containing monooxygenase form 3 (FMO3) enzyme in an individual, detecting a susceptibility of an individual to a substrate of the FMO3 enzyme in an individual, and for detecting a predisposition of an individual to a disorder associated with an exposure to a heteroatom-containing chemical compound, by detecting mutations or polymorphic variants of the FMO3 gene.